



CASK gene

calcium/calmodulin dependent serine protein kinase

Normal Function

The CASK gene provides instructions for making a protein called calcium/calmodulin-dependent serine protein kinase (CASK). The CASK protein is primarily found in nerve cells (neurons) in the brain, where it helps control the activity (expression) of other genes that are involved in brain development. It also helps regulate the movement of chemicals called neurotransmitters and of charged atoms (ions), which are necessary for signaling between neurons. Research suggests that the CASK protein may also interact with the protein produced from another gene, *FRMD7*, to promote development of the nerves that control eye movement (the oculomotor neural network).

Health Conditions Related to Genetic Changes

CASK-related intellectual disability

More than 35 CASK gene mutations have been identified in people with CASK-related intellectual disability. This disorder affects brain development and has two main forms: a severe form called microcephaly with pontine and cerebellar hypoplasia (MICPCH), and a milder form called X-linked intellectual disability (XL-ID) with or without nystagmus.

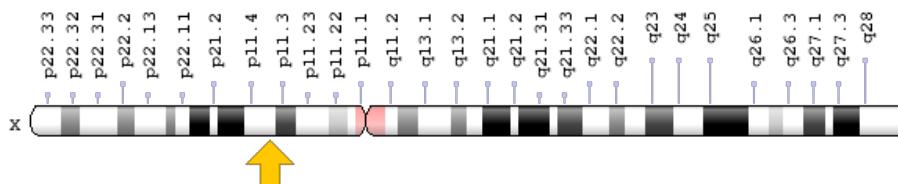
The mutations that cause CASK-related intellectual disability affect the role of the CASK protein in brain development and function. MICPCH is caused by mutations that eliminate CASK function, while mutations that impair the function of this protein cause XL-ID with or without nystagmus. Nystagmus refers to rapid, involuntary back-and-forth eye movements. Affected individuals with nystagmus may have CASK gene mutations that disrupt the interaction between the CASK protein and the protein produced from the *FRMD7* gene, leading to problems with the development of the oculomotor neural network and resulting in abnormal eye movements.

FG syndrome

Chromosomal Location

Cytogenetic Location: Xp11.4, which is the short (p) arm of the X chromosome at position 11.4

Molecular Location: base pairs 41,514,934 to 41,923,525 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- calcium/calmodulin-dependent serine protein kinase (MAGUK family)
- CAMGUK
- CMG
- CSKP_HUMAN
- hCASK
- LIN2
- protein lin-2 homolog
- TNRC8

Additional Information & Resources

GeneReviews

- CASK-Related Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK169825>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CASK%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE
<http://omim.org/entry/300172>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CASK.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CASK%5Bgene%5D>
- HGNC Gene Family: Membrane associated guanylate kinases
<http://www.genenames.org/cgi-bin/genefamilies/set/904>
- HGNC Gene Family: PDZ domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1220>
- HGNC Gene Family: Trinucleotide repeat containing
<http://www.genenames.org/cgi-bin/genefamilies/set/775>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1497
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8573>
- UniProt
<http://www.uniprot.org/uniprot/O14936>

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